



X-linked sideroblastic anemia and ataxia

X-linked sideroblastic anemia and ataxia is a rare condition characterized by a blood disorder called sideroblastic anemia and movement problems known as ataxia. This condition occurs only in males.

Sideroblastic anemia results when developing red blood cells called erythroblasts do not make enough hemoglobin, which is the protein that carries oxygen in the blood. People with X-linked sideroblastic anemia and ataxia have mature red blood cells that are smaller than normal (microcytic) and appear pale (hypochromic) because of the shortage of hemoglobin. This disorder also leads to an abnormal accumulation of iron in red blood cells. The iron-loaded erythroblasts, which are present in bone marrow, are called ring sideroblasts. These abnormal cells give the condition its name. Unlike other forms of sideroblastic anemia, X-linked sideroblastic anemia and ataxia does not cause a potentially dangerous buildup of iron in the body. The anemia is typically mild and usually does not cause any symptoms.

X-linked sideroblastic anemia and ataxia causes problems with balance and coordination that appear early in life. The ataxia primarily affects the trunk, making it difficult to sit, stand, and walk unassisted. In addition to ataxia, people with this condition often have trouble coordinating movements that involve judging distance or scale (dysmetria) and find it difficult to make rapid, alternating movements (dysdiadochokinesis). Mild speech difficulties (dysarthria), tremor, and abnormal eye movements have also been reported in some affected individuals.

Frequency

X-linked sideroblastic anemia and ataxia is a rare disorder; only a few affected families have been reported.

Genetic Changes

Mutations in the *ABCB7* gene cause X-linked sideroblastic anemia and ataxia. The *ABCB7* gene provides instructions for making a protein that is critical for heme production. Heme is a component of the hemoglobin protein, which is vital for supplying oxygen to the entire body. The *ABCB7* protein also plays a role in the formation of certain proteins containing clusters of iron and sulfur atoms. Overall, researchers believe that the *ABCB7* protein helps maintain an appropriate balance of iron (iron homeostasis) in developing red blood cells.

ABCB7 mutations slightly alter the structure of the *ABCB7* protein, disrupting its usual role in heme production and iron homeostasis. Anemia results when heme cannot be

produced normally, and therefore not enough hemoglobin is made. It is unclear how changes in the *ABCB7* gene lead to ataxia and other problems with movement.

Inheritance Pattern

This condition is inherited in an X-linked recessive pattern. The gene associated with this condition is located on the X chromosome, which is one of the two sex chromosomes. In males (who have only one X chromosome), one altered copy of the gene in each cell is sufficient to cause the condition. In females (who have two X chromosomes), a mutation would have to occur in both copies of the gene to cause the disorder. Because it is unlikely that females will have two altered copies of this gene, males are affected by X-linked recessive disorders much more frequently than females. A characteristic of X-linked inheritance is that fathers cannot pass X-linked traits to their sons.

In X-linked recessive inheritance, a female with one altered copy of the gene in each cell is called a carrier. Carriers of an *ABCB7* mutation can pass on the mutated gene but do not develop ataxia or other health problems associated with X-linked sideroblastic anemia and ataxia. However, carriers may have abnormally small, pale red blood cells and related changes that can be detected with a blood test.

Other Names for This Condition

- XLSA/A

Diagnosis & Management

Genetic Testing

- Genetic Testing Registry: Anemia sideroblastic and spinocerebellar ataxia
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C1845028/>

Other Diagnosis and Management Resources

- GeneReview: X-Linked Sideroblastic Anemia and Ataxia
<https://www.ncbi.nlm.nih.gov/books/NBK1321>
- MedlinePlus Encyclopedia: Anemia
<https://medlineplus.gov/ency/article/000560.htm>

General Information from MedlinePlus

- Diagnostic Tests
<https://medlineplus.gov/diagnostictests.html>
- Drug Therapy
<https://medlineplus.gov/drugtherapy.html>
- Genetic Counseling
<https://medlineplus.gov/geneticcounseling.html>

- Palliative Care
<https://medlineplus.gov/palliativecare.html>
- Surgery and Rehabilitation
<https://medlineplus.gov/surgeryandrehabilitation.html>

Additional Information & Resources

MedlinePlus

- Encyclopedia: Anemia
<https://medlineplus.gov/ency/article/000560.htm>
- Health Topic: Anemia
<https://medlineplus.gov/anemia.html>
- Health Topic: Cerebellar Disorders
<https://medlineplus.gov/cerebellardisorders.html>

Genetic and Rare Diseases Information Center

- Anemia sideroblastic and spinocerebellar ataxia
<https://rarediseases.info.nih.gov/diseases/668/anemia-sideroblastic-and-spinocerebellar-ataxia>

Additional NIH Resources

- National Institute of Neurological Disorders and Stroke: Ataxias and Cerebellar or Spinocerebellar Degeneration
<https://www.ninds.nih.gov/Disorders/All-Disorders/Ataxias-and-Cerebellar-or-Spinocerebellar-Degeneration-Information-Page>

Educational Resources

- MalaCards: x-linked sideroblastic anemia with ataxia
http://www.malacards.org/card/x_linked_sideroblastic_anemia_with_ataxia
- Orphanet: X-linked sideroblastic anemia and ataxia
http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=2802

Patient Support and Advocacy Resources

- National Ataxia Foundation
<http://www.ataxia.org/>
- National Organization for Rare Disorders (NORD): Sideroblastic anemias
<https://rarediseases.org/rare-diseases/anemias-sideroblastic/>
- Resource list from the University of Kansas Medical Center: Ataxia
<http://www.kumc.edu/gec/support/ataxia.html>

GeneReviews

- X-Linked Sideroblastic Anemia and Ataxia
<https://www.ncbi.nlm.nih.gov/books/NBK1321>

ClinicalTrials.gov

- ClinicalTrials.gov
<https://clinicaltrials.gov/ct2/results?cond=%22X-linked+sideroblastic+anemia+and+ataxia%22+OR+%22Sideroblastic+Anemia%22>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28x-linked%5BTIAB%5D+AND+sideroblastic+anemia%5BTIAB%5D+AND+ataxia%5BTIAB%5D%29+OR+%28xlsa/a%5BTIAB%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D>

OMIM

- ANEMIA, SIDEROBLASTIC, AND SPINOCEREBELLAR ATAXIA
<http://omim.org/entry/301310>

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